

### **Abstract**

The present invention relates to nucleic acid molecules, preferably genomic sequences, encoding an ATP-gated ion channel P2X7R which contain a mutation in the 5'UTR or 3'UTR regions, a mutation in exon 3, 5, 6, 8 or 13 or in introns 1, 3, 4, 5, 6, 7, 9, 11 or 12 or a deletion in exon 13, which allow to diagnose affective disorders. The invention further relates to polypeptides encoded by said nucleic acid molecules vectors and host cells comprising said nucleic acid molecules as well as to methods for producing polypeptides encoded by said nucleic acid molecules. The present invention also provides antibodies specifically directed to polypeptides encoded by said nucleic acid molecules and aptamers specifically binding said nucleic acid molecules.

Additionally, primers for selectively amplifying said nucleic acid molecules are provided in the present invention as well as kits, compositions, particularly pharmaceutical and diagnostic compositions comprising said nucleic acid molecules, vectors, polypeptides, aptamers, antibodies and/or primers. Moreover, the present invention relates to methods for diagnosing affective disorders associated with a non-functional P2X7R protein, an altered ATP-gating of the P2X7R protein, an over- or underexpression of the P2X7R protein or associated with the presence of any one of the aforementioned nucleic acid molecules or polypeptides encoded thereby. Additionally, the present invention relates to uses and methods for treating affective disorders employing a functional or non-functional ATP-gated ion-channel P2X7R.

The present invention also relates to uses of modulators of P2X7R activity for treating affective diseases.

Furthermore, the present invention also relates to methods for identifying and characterizing compounds which are capable of specifically interacting with or altering the characteristics of the polypeptides of the present invention as well as to methods for the production of pharmaceutical compositions.